



COLEC10 gene

collectin subfamily member 10

Normal Function

The *COLEC10* gene provides instructions for making a protein called collectin liver 1 (CL-L1). This protein is involved in a series of steps called the lectin complement pathway, which is thought to help direct the movement (migration) of cells during development before birth to form the organs and systems of the body. The lectin complement pathway appears to be particularly important in directing the migration of neural crest cells. These cells give rise to various tissues including many tissues in the face and skull, glands that produce hormones (endocrine glands), and portions of the nervous system. After birth, the lectin complement pathway is involved in the immune system.

Health Conditions Related to Genetic Changes

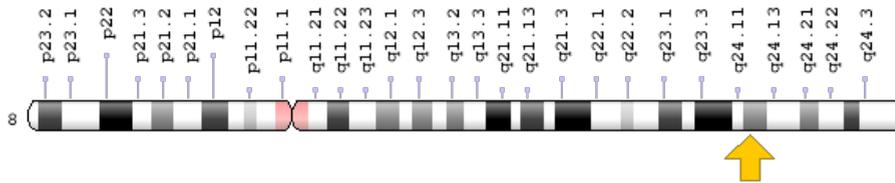
3MC syndrome

At least three *COLEC10* gene mutations have been identified in people with 3MC syndrome, a disorder characterized by unusual facial features and a variety of problems affecting other tissues and organs. The *COLEC10* gene mutations that cause 3MC syndrome impair or eliminate the function of the CL-L1 protein, resulting in faulty control of cell migration in early development and leading to the various abnormalities that occur in this disorder. Researchers suggest that similar pathways in the immune system can compensate for problems in the lectin complement pathway, which explains why immune system abnormalities are not part of 3MC syndrome.

Chromosomal Location

Cytogenetic Location: 8q24.12, which is the long (q) arm of chromosome 8 at position 24.12

Molecular Location: base pairs 118,952,233 to 119,108,455 on chromosome 8 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3MC3
- CL-34
- CL-L1
- CLL1
- collectin-10 isoform 1 precursor
- collectin-10 isoform 2
- collectin-34
- collectin liver 1
- collectin liver protein 1
- collectin sub-family member 10 (C-type lectin)

Additional Information & Resources

Educational Resources

- Immunobiology --The Immune System in Health and Disease (fifth edition, 2001):
The Mannan-binding Lectin Pathway is Homologous to the Classical Pathway
<https://www.ncbi.nlm.nih.gov/books/NBK27100/#A170>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28COLEC10%5BTIAB%5D%29+OR+%28collectin+subfamily+member+10%5BTIAB%5D%29%29+OR+%28%28CL-34%5BTIAB%5D%29+OR+%28CL-L1%5BTIAB%5D%29+OR+%28CLL1%5BTIAB%5D%29+OR+%28collectin+liver+1%5BTIAB%5D%29+OR+%28collectin+liver+protein+1%5BTIAB%5D%29+OR+%28collectin+sub-family+member+10%5BTIAB%5D%29+OR+%28collectin-10+isoform+1+precursor%5BTIAB%5D%29+OR+%28collectin-10+isoform+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- COLLECTIN 10
<http://omim.org/entry/607620>

Research Resources

- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2220
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:10584>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10584>
- UniProt
<https://www.uniprot.org/uniprot/Q9Y6Z7>

Sources for This Summary

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